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## **Clinico-epidemiological profile of congenital ocular anomalies in Western Odisha: A hospital based cross sectional study**

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**Abstract**---Congenital birth defects are major cause of poor health among infants affecting their survivability. Although such anomalies of the eyes are uncommon, the impact they have on the quality of life are more than significant. This study aimed to describe the clinical profile of congenital ocular anomalies with various epidemiological parameters in a tertiary health care center in western Orissa. The study screened 3674 patients, in the age group of 0 – 14 years, who were delivered or attended the out-patient Department V.S.S. Medical College, Burla, Odisha and included 151 eyes of 99 cases of various types of congenital anomalies. Detailed history and other necessary data were collected by trained ophthalmologists and descriptive analyses performed. The incidence of congenital ocular anomalies was found to be 2.69%. Nasolacrimal duct anomalies (47 eyes) were the most frequently occurring congenital ocular anomaly (31.1%); followed by congenital cataract (41 cases; 27.3%) and coloboma eye (18 cases; 11.9%). There was a slight male preponderance, constituting 56.57% of the total cases. Majority of the patients were less than 5 years old (68 %) and the incidence of congenital ocular anomalies to be highest

(68.69%) in the age group of 0 – 5 years. Single eye involvement was seen in 47 cases (47.47%), whereas bilateral involvement was seen in 52 (52.53%) cases. History of consanguinity was found to be present in 13.13% of cases, with increased risk of bilateral involvement (84.6% cases). The most common presenting symptom was defective vision (38.4%), followed by watering from eyes (32.3%) and white pupillary reflex (14.1%). A total of 60 eyes (39.7%) required surgery. Congenital ocular anomalies are a common cause of ocular morbidity, comprising 2.7% of total cases in the paediatric age group. Early diagnosis, referral and appropriate intervention would help prevent majority of permanent sequelae in congenital ocular anomalies.

**Keywords**---Congenital anomalies, cataract, nasolacrimal duct obstruction, children, ocular disorders.

## **Introduction**

Congenital birth defects are major cause of poor health among infants affecting their survivability. Although such anomalies of the eyes are uncommon, the impact they have on the quality of life are more than significant. By definition, congenital anomalies include those deformities that occur during embryologic or antenatal development. Congenital ocular anomalies are major contributors to childhood visual morbidity. These disorders affect the disadvantaged populations disproportionately with poorer access to preventive, screening and curative opportunities.

Epidemiological estimates place an incidence rate of congenital eye disorders at 3.68/10,000 newborns, and of eye malformations are associated with other congenital abnormalities. (1) The commonest among these congenital ocular disorders are buphthalmos (38%), cataracts (35%), and nasolacrimal duct obstruction (14%). (2) Almost 86% of these patients need corrective surgery. (3) Other less frequent anomalies are anophthalmia/microphthalmia, limbal dermoid cysts and aniridia. (4)

Certain anomalies result from direct damage to the ocular or adnexal tissues during their embryologic development, but many ophthalmologic signs and symptoms occur as early or late complication of the disturbed anatomy in the surrounding structures. While these conditions are genetically transmitted, very few risk factors and pre-natal screening methods have been identified. Consanguinity of marriage and previous history of defects in siblings are some strong correlates of congenital eye disorders. Other possible etiological factors, especially for congenital cataracts have been identified such as antenatal maternal infections and malnutrition. Most important among them is Congenital rubella Syndrome. (5) Another common risk factor associated is maternal vitamin-A deficiency. (6–8)

Besides the common disorders, other isolated ocular anomalies are also seen that are associated with systemic developmental anomalies as a part of various clinical syndromes. Estimates of the incidence of congenital anomalies are required for

planning and prioritizing preventive programs. Such incidence data are currently not available in many settings. With this background, this study is planned with an objective of describing the clinical profile of congenital ocular anomalies with various epidemiological parameters in a tertiary health care center in western Orissa.

## **Methodology**

This was a hospital based cross sectional study conducted at the department of Ophthalmology at VSS Medical College and Hospital, Burla in Odisha. The study participants were recruited from the patients attending the outpatient departments with any apparent congenital ophthalmic disorder, during a period of 2 years. We used consecutive non-random sampling to select participants due to the rare nature of the conditions under study.

The neonates who were delivered at the same hospital were also included in the study after a systematic screening. We included all children, below 14 years of age, who had a newly or previously diagnosed congenital anomaly, irrespective of treatment undertaken. Children who died after diagnosis were also included. Patients with unreliable history, improper or inadequate record of diagnosis or no follow up, or those who died before diagnosis were excluded from our study. We also excluded those patients among whom a cause could not be ascertained to be due to congenital cause, i.e., any developmental or acquired cases.

A detailed history of the patient was collected using a predefined form that included demographic characteristics, clinical examination, risk factor assessment, maternal obstetric history – any significant antenatal events, any prevailing chronic disease process, any systemic or infectious disease affecting during the antepartum period, any history of drug intake or ingestion of toxic substances, maternal malnutrition, any food supplementation prescribed and/or taken or relevant family history.

The clinical examination included the following parameters performed using standard procedure followed at the host institution:

- Visual acuity (Snellen's chart) – to correlate the degree of severity and assess prognosis,
- Slit lamp examination – a detailed examination of the anterior segment and anterior hyaloid face to confirm any morphological or structural anomalies,
- L.P.I. test – to confirm for any nasolacrimal duct obstruction,
- I.O.P. measurement by applanation / Schiottz tonometry – to diagnose primary congenital glaucoma cases,
- Keratometry, A-scan ultrasonography – to measure corneal diameter and ascertain power of intraocular lens to be implanted in congenital cataract cases,
- Ophthalmoscopy (both direct and indirect) – to rule out any posterior segment anomalies.

The patients were managed according to the standard treatment protocols of the institute, including corrective surgery if necessary. Data were collected by trained

Ophthalmology residents. All data was tabulated in an excel form and analysed using SPSS software. Descriptive summaries were reported using frequencies and proportions. The study received ethical approval from the Institutional ethical clearance committee of host institute and informed consent was obtained from all the parents/guardians of the participating children.

## Results

Our study reports findings from 99 cases of congenital ocular anomalies (151 eyes) who were included in the 2 year study period. These were selected by screening from 3674 patients resulting in an incidence rate of 2.69%. Out of total 99 patients, 56 (56.57%) cases were males and 43 (43.43%) cases were females, with a male:female ratio of 1.3:1. Among them, right and left eyes were involved individually in 29 (29.29%) and 18 (18.18%) cases respectively, with a collective total of 47 cases (47.47%), whereas *bilateral involvement* was seen in 52 (52.53%) cases.

In our study, congenital nasolacrimal duct obstruction, CNLDO, (47 cases; 31.1%) was found to be the most common type of congenital ocular anomaly in this setup, followed by congenital cataract (41 cases; 27.3%), and coloboma of eye (18 cases; 11.9%). The other conditions are shown below in table-1.

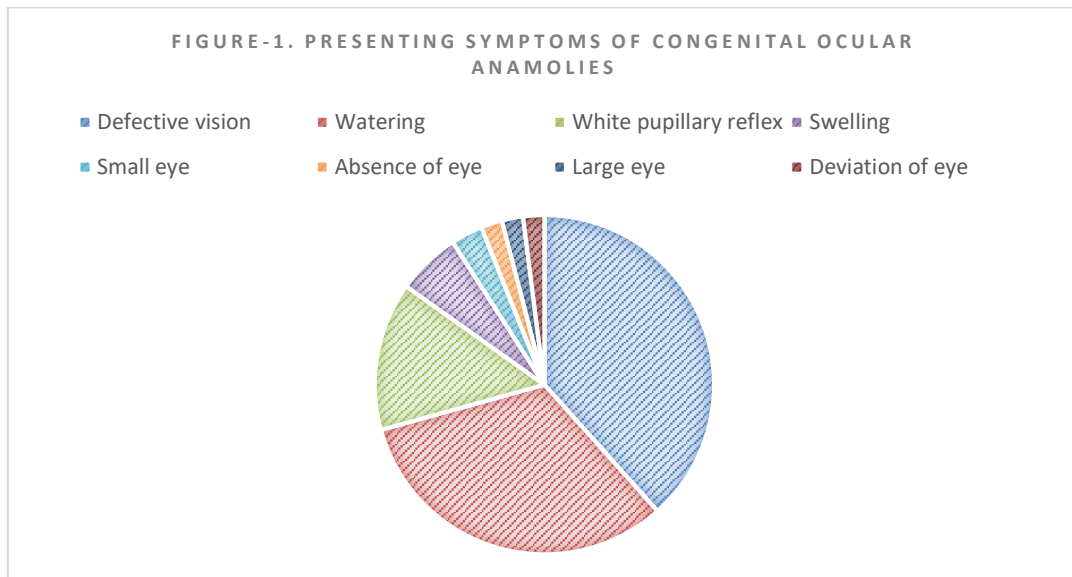
Sl. No.	Congenital ocular disorder	No. of cases	No. of eyes affected	% of eyes affected
1.	<i>Congenital Naso-Lacrimal Duct Obstruction</i>	32	47	31.1%
2.	<i>Congenital Cataract</i>	26	41	27.3%
3.	<i>Coloboma</i>	11	18	11.9%
4.	<i>Anophthalmia / Microphthalmia</i>	08	13	8.6%
5.	<i>Persistent Pupillary Membrane</i>	04	07	4.7%
6.	<i>Limbal Dermoid / Dermoid Cyst</i>	05	05	3.4%
7.	<i>Medullated Nerve Fibre</i>	04	05	3.4%
8.	<i>Heterochromia Iridis</i>	02	04	2.8%
9.	<i>Congenital Glaucoma</i>	02	04	2.8%
10.	<i>Aniridia</i>	02	03	2.0%
11.	<i>Congenital Esotropia</i>	02	02	1.3%
12.	<i>Lipoma of Lid</i>	01	01	0.7%
	TOTAL	99	151	100%

The incidence of congenital ocular anomalies was found to be highest (68.69%) in the age group of 0 – 5 years, and gradually decreases in the further age groups. History of consanguineous marriage was found in the parents of 13 (13.13%)

cases. Out of 13 cases, 11 (84.62%) cases were found to have bilateral involvement of eyes.

Among the study participants, 90 cases had an uneventful antepartum period. Maternal malnutrition was associated with 2 cases of coloboma. History of infection with fever at 5 months gestation was seen in a case of anophthalmos (OD) and use of antimalarial drug history was found in a case of congenital cataract (OU) with microcornea (OD). History of flu-like symptoms at 4 months gestation was seen in a case of congenital cataract (OU). History of abortion of previous child was found in 2 cases of coloboma iris (OU). History of attempted abortion at 2 ½ months gestation was given in a case of anophthalmos (OS) with microphthalmos (OD), whereas history of threatened abortion was seen in a case of anophthalmos (OS) with microphthalmos (OD).

In our study, the most common mode of presenting symptom was defective vision (38.39%), followed by watering from eyes (32.32%) and white pupillary reflex (14.14%) as shown in figure-1 below.



Associated systemic defects were seen in 6 cases. Limb defects and albinism are found in 2 cases each. Congenital heart defect was found in 1 case of coloboma iris and choroid (OU), and pigeon-chest deformity with CVS defects was seen in 1 case of congenital cataract (OD).

Congenital cataract was found to be equal in distribution in both the sexes. But CNLDO was present in a higher proportion in males (22 cases) as compared in females (10 cases). Cases of coloboma were also found to be more in males (7 cases) than females (4 cases). There was no sex predilection in cases of microphthalmia / anophthalmia.

Visual acuity could not be recorded in sixty five cases due to non-compliance of the patients. Nineteen cases were found to have visual acuity better than 6/12,

followed by 6/18 – 6/36 group (ten cases) and >6/60 group (five cases). In our study, 60 eyes (39.7%) were undertaken for surgeries as detailed below in table-2.

Ocular anomaly in which surgery performed	No. of cases (eyes) of ocular anomaly	No. of eyes in which surgery performed	% of eyes in which surgery performed
Congenital cataract	26 (41)	32	78.0%
CNLDO	36 (47)	18	38.3%
Congenital glaucoma	02 (04)	04	100%
Limbal dermoid / Dermoid cyst	05 (05)	05	100%
Congenital esotropia	02 (02)	01	50.0%
TOTAL	99 (151)	60	39.7%

Out of 41 cases of congenital cataract, cataractous lens extraction followed by lens implantation was performed in 32 cases. Majority of cases of congenital nasolacrimal duct obstruction responded to massaging of the lacrimal sac area and maintenance of proper lid hygiene. 14 cases were cured by first probing and another 2 cases required a second probing. In 4 cases, dacryocystorhinostomy was performed followed by relieving of obstruction of the obstruction. All the cases of primary corneal glaucoma required surgery. In 2 cases, goniotomy was performed, and in the remaining 2 cases, trabeculotomy was required. *In toto* excision was required in all the cases of limbal dermoids / dermoid cysts followed by normal healing process.

## Discussion

This was a hospital based study on 3674 patients in the age group of 0 – 14 years in a tertiary health care centre over a period of 2 years. A total of 99 cases of congenital ocular anomalies were found that included 151 eyes. A detailed analysis was done to understand the incidence and types of such anomalies, age and sex distribution, involvement of single or both eyes, modes of presentation, visual status at presentation, associated ocular / systemic findings and their modes of management.

We report an incidence of congenital ocular anomalies to be 2.69% in all the patients presenting in the 0 -14 years age group. This is significantly higher as compared to other studies conducted in similar low resource settings across the world. (3) However, some studies in high income settings such as Spain found a higher incidence of 3.68/10000 newborns as compared to our study. (1)

The most common disorder found in our study was CNLDO. Internationally, nasolacrimal obstruction is known to occur in 2-6% of newborns. (9,10) The second most common anomaly found was congenital cataract. Others have reported an incidence of congenital cataract to be between 1-6 per100,000 live births. (11) Congenital cataracts are seen more in the developing countries like India as compared to high income societies by a factor of ten times. (12)

We found a slight male preponderance and the male:female ratio was 1.3:1. Similar findings were reported by Lawan et.al. but at a ratio of 2.3:1. (3) However, in a retrospective non-comparative case-series study of 54 cases by Chuka-Okosa *et. al.* in 2005, the male:female ratio was found to be 1:1.2. (2) We found an equal in distribution of congenital cataract or microphthalmia / anophthalmia in both the sexes which has been corroborated by previous authors. (6,13) The small number of samples included in all these studies make any comparisons regarding gender distributions needless.

We found the incidence of congenital ocular anomalies to be highest (68.69%) in the age group of 0 – 5 years. The adjusted age-specific incidence of new diagnosis of congenital and infantile cataract in the first year of life is estimated to be 2.49 per 10,000 children, that increases to 3.18 per 10,000 at 5 years and 3.46 per 10,000 by 15 years. (6)

Our study found that history of consanguineous marriage of parents was there in 13.13% of cases. Other antenatal risk factors reported were malnutrition, maternal infection, bad maternal obstetric history. Consanguinity has been reported as a risk factor in other studies as well, but at a much higher risk attached to it. (14) Similarly, one or more of the four infectious agents of TORCH infection were detected in 20.2 per cent of the study population in a study from India. (15) The most common eye defects attributed to maternal rubella, in descending order of frequency, are congenital cataract, chorioretinitis, microphthalmos, and glaucoma. (8,16)

The most common mode of presenting symptom was defective vision (38.39%), followed by watering from eyes (32.32%) in our study. In CNLDO cases, the most common presenting symptom is watering from eyes and similar findings have been reported by Mounir et.al. (9) In cases of congenital cataract, the most common presenting symptom was leucocoria (white pupillary reflex) and this was also corroborated by multiple previous studies. (17,18)

We found that congenital systemic associations are present in 6% of cases. While most other studies have found similar associations, the extent of co-occurrence reported is generally higher ranging to about 20% depending on the type of anomaly. (1,9) In our study, around 40% patients needed surgery. This included 78% of cases of congenital cataract, 38.3% cases of CNLDO and all the cases of primary corneal glaucoma or limbal dermoids/dermoid cysts who required surgery. Congenital eye and adnexal anomalies accounted for between 25-50% of eye surgeries in children and the success rates are generally good. (3,17,19)

The study is one of few from India reporting congenital eye anomalies in children. As it is an observational study and as the sample size is low, the findings could not be generalized to the larger population and this is a limitation of our study. In conclusion, congenital ocular anomalies are a common cause of ocular morbidity, comprising 2.7% of total cases in the pediatric age group. Majority of the cases have been reported in the age group of 0 – 5 years of age and congenital nasolacrimal duct obstruction was the most common congenital ocular anomaly present in children followed by congenital cataract. Most of the cases were treatable, especially CNLDO and congenital cataract. Early diagnosis, prompt

reference from community levels and appropriate intervention would help prevent majority of permanent sequelae in congenital ocular anomalies.

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